Book Reviews

Birth Defects: Original Article Series, V, 3. First Conference on The Clinical Delineation of Birth Defects, Part III, 'Limb Malformations'. (Pp. 234; figures + tables. £7.) New York: The National Foundation—March of Dimes. Edinburgh: E. and S. Livingstone for Williams and Wilkins.

This is a report of the proceedings at the First conference on the 'Clinical Delineation of Birth Defects' held at the Johns Hopkins Hospital, Baltimore in May 1968. Volume III is devoted to limb malformations and attempts to distinguish fundamentally distinct anomalies or groups of anomalies as a first step in the elucidation of pathogenesis, and as a guide to genetic counselling and planning of possible future treatment.

A review of the whole problem with particular reference to the classification of anomalies was given by Dr Lenz, Dr Freire-Maia, and by Dr O'Rahilly. Dr Pfeiffer reviewed the numerous 'head and hand' anomalies and dermatoglyphs were dealt with by Dr Alter and Dr Warburton. Much of the volume deals with a variety of rare syndromes, some never previously reported, and there are a large number of excellent illustrations throughout.

The largest single contribution is from Dr Temtamy and Dr McKusick who are to be congratulated on their classification of limb anomalies based on anatomical and genetic characteristics. This was arrived at by a careful study of many families seen personally and appraisal of other reported cases. They take each type of deformity and consider it first as an isolated phenomenon and then where it occurs as part of a generalized disorder or syndrome. This is a more helpful way of considering the numerous malformations of the limbs than those classifications based solely on details of anatomical variation.

The anomalies dealt with are still only at the stage of clinical description and genetic analysis and the book will be mainly of value to clinicians and clinical geneticists. It is to be welcomed for its presentation of carefully observed cases and it is to be hoped that there will be future conferences where material collected from many centres can be presented and compared in this way.

RUTH WYNNE-DAVIES

Birth Defects: Original Article Series, V, 2. First Conference on The Clinical Delineation of Birth Defects, Part II 'Malformation Syndromes'. (Pp. 284; figures + tables. £7.) New York: The National Foundation—March of Dimes. Edinburgh: E. and S. Livingstone for Williams and Wilkins.

The first conference of the 'Clinical Delineation of Birth Defects' was held over 5 days in May 1968 at the Johns Hopkins Hospital and proceedings have now been published in 5 volumes, of which this is the second. It is aptly placed between Part I, a thin volume of three essays on biological development and nosology of genetic disease and Part III which is made up of a series of articles on limb malformations. Part IV was devoted to skeletal dysplasias and Part V to the phenotypic aspects of chromosomal aberrations.

The prologue to this volume by Dr Opitz clearly underlines the difficulties that face those who study and try to classify the malformation syndromes. This introduces a series of articles on what may be specific malformation syndromes. Some of these are reviews; Rubinstein's paper on the broad thumb and big toe syndrome is one excellent example. Others are restricted to the description of very rare and familial conditions. In all, 19 conditions are covered. Many of these articles are written by the authorities on the subject though the Wisconsin group, headed by Dr Opitz, appears to overwhelm the others. This volume is not aimed at giving a comprehensive list and description of all the malformation syndromes, so it should not be bought in the hope that it will provide this information. It is perhaps unfortunate that the Wisconsin group have seen fit to use the initial letter of the index family as titles for some of the conditions they describe, and although this should be considered a brave attempt to avoid eponyms single or even multiple letters are not impressive as *aide memoirs* and are certainly not descriptive. The articles themselves are clear and are beautifully illustrated with numerous photographs. There is in addition a small number of articles which consider malformations on a rather broader basis; Gorlin describes 8 specific syndromes that affect the face and George Fraser considers malformations with eye and ear involvement. Pruzansky shows that careful *x*-ray measurements of the developing mandible gives precision to diagnosis and provides a useful record which will define the degree of defect associated with a specific condition. A further one sixth of the book is filled with straight case reports that are without comment.

The volume is almost closed by an epilogue by Polani who reminds us that disorders that bear a resemblance to each other can only be effectively separated from each other when linked to some recognizable metabolic or cytological label. Right at the end David W. Smith sneaks in with an appendix of simple tables that attempt to classify abnormalities according to the most obvious affected tissue or organ or to the underlying defect when it is known.

This is a book which clinical geneticists and those interested in malformations will enjoy looking at and reading. It is probably best bought together with the companion volumes rather than in isolation. It supplements but does not replace other illustrated catalogues which have been recently published on the malformation syndromes.

J. INSLEY

Alzheimer's Disease and Related Conditions. A Ciba Foundation Symposium. Edited by G. E. W. Wolstenholme and Maeve O'Connor. (Pp. xi+316; figures+tables. £4.) London: J. and A. Churchill. 1970.

Alzheimer's disease is a moderately infrequent form of presenile dementia, with a well-defined course and clinical picture, and characteristic cerebral pathology. Genetically it is interesting because of the indications of genetic heterogeneity within this clinico-pathologically delimited syndrome. A number of families have been described, in his contribution to the symposium Pratt says 'more than a dozen', in which a completely typical form of autosomal dominant inheritance is shown, with equal sex representation, and nearly complete penetrance. In comprehensive series, however, selected on a clinico-pathological basis, these typical families do not appear; and what one sees instead is a moderately raised familial incidence and a female preponderance of about 3:1, ie, the indications of a polygenic genetical basis. The genetical evidence, therefore, is strongly in favour of regarding the syndrome as being a mixture of at least two different and independent conditions. This is a conclusion which, for the most part, both clinicians and pathologists are unwilling to accept, as is made plain in these papers.

The symposium reported here was predominantly a pathological one, with interesting and original pathological work. Participants came from Belgium, Germany, the Argentine, Sweden, Hungary, the USA (7) and Britain (10). Active work is going on in histology, histochemistry, and electron-microscopy. Most promising, perhaps, is the study of the ultramicroscopic structure, ie the abnormal intracellular microtubules, which differentiate more than one kind of lesion, and may lead to a successful breaking down of the clinico-pathological continuum extending into normal senescence, senile dementia, and cerebrovascular disease, by which most workers are at present both obsessed and bewildered.

The reviewer would suppose that this book is an important one for the neuropathologist. Its genetical interest is not great. Apart from Pratt's paper reviewing the present state of genetical understanding, the main interest lies in the communication by Jacob (Marburg, Germany) of a family with 6 females in 3 generations showing a form of Alzheimer's disease with, in every case, the unusual feature of myoclonic twitchings appearing early in the course. The literature is reviewed by Jacob.

The book is well produced, with an abundance of good pathological illustrations, good papers, lively discussions, but rather inconclusive results.

E. SLATER

Serendipity in St. Helena. A Genetical and Medical Study of an Isolated Community. By Ian Shine. (Pp. xv+187; illustrated+tables. £3·75.) Oxford: Pergamon. 1970.

St. Helena shares with its near neighbour Tristan da Cuhna many of the aspects of an isolated community. It is however an older settlement and has population of some 5000 against the 350 or so of Tristan da Cuhna. A genetical survey of this highly inbred population with a culture of its own and its particular racial amalgam is a worth-while task which Dr. Ian Shine carried out single-handed while acting as medical officer to the island during his national service in the Army. The present book, based on an examination of almost the whole of the population, records his findings.

One study was devoted to hallux valgus. On St. Helena as elsewhere it was common among those who wear shoes and uncommon among the barefooted. Regression analysis showed however that an additional factor, probably genetic, was also operative. Furthermore some families seemed particularly susceptible to the deforming effect of shoes, while others developed the deformity in spite of their barefootedness. An equally detailed study on the frequency of ischaemic heart disease revealed a substantial incidence in the 1500 subjects over the age of 30 examined. This was unexpected, but clear aetiological conclusions were not possible.

In the course of the overall survey a large number of genetic disorders were observed, including some that are distinctly uncommon, such as albinism, retinitis pigmentosa, lithopaedion, icthyosiform erythrodermia and Christmas disease. Undescribed disorders such as gross unilateral genu valgum, 'familial St. Helenian fever', and dwarfs with pseudobrachydactyly constituted a group of considerable interest. A high incidence of pterygium presented a puzzle, for the commonly accepted exciting factors, such as dust, wind, and dry air, could not be invoked as an explanation.

The genetic problems disclosed in this investigation are discussed in a critical penultimate chapter on inbreeding. Like the rest of the text this is competent, but this useful volume is not well served by its rather flippant title and somewhat jaunty style.

ARNOLD SORSBY

The De Lange Syndrome. By J. M. Berg, B. D. McCreary, M. A. C. Ridler, and G. F. Smith. (Pp. vii+127; figures+tables. £3·15; $8.40.) Oxford: Pergamon Press. 1970.

The de Lange syndrome was first described by a Dutch paediatrician from Amsterdam in 1933. Since that time a wealth of data has accumulated on the clinical